FILE SEGMENT: 022 Human Genetics
014 Radiology
033 Orthopedic Surgery
LANGUAGE: English
AB The Winchester syndrome is characterized by dwarfism, abnormal facies, contractures, corneal opacities, osteoporosis, dissolution of carpal and tarsal bones and intra and periarticular joint destruction simulating severe rheumatoid arthritis. Clinical, radiographic, pathologic and ultrastructural studies were performed on two affected sibs of a consanguineous Mexican marriage. Carpal tarsal osteolysis eventuates in nearly complete loss of these bones and marked medial hypertrophy of arterioles was found in the dense fibrous tissue which replaces the carpal bones. Excessive resorption of histochemically abnormal bone is evident on bone biopsy. Fibroblastic proliferation in the deep dermis occurs in widespread areas. Electron microscopic studies of skin fibroblasts reveal swelling and degeneration of mitochondria and dilation of endoplasmic reticulum. No lysosomal accumulation was observed in skin, gingiva, bone marrow or cartilage indicating that the Winchester syndrome is a nonlysosomal connective tissue disease, and should be removed from the list of mucopolysaccharidoses. This syndrome appears to be inherited as an autosomal recessive trait.

L11 ANSWER 8 OF 8 EMBASE
ACCESSION NUMBER: 75021070 EMBASE
DOCUMENT NUMBER: 1975021070
TITLE: The Winchester syndrome: a nonlysosomal connective tissue disease.
AUTHOR: Hollister D.W.; Rimoin D.L.; Lachman R.S.; et al.
CORPORATE SOURCE: Div. Med. Genet., Dept. Ped. Med., Harbor Gen. Hosp. UCLA Sch. Med., Torrance, Calif. 90509, United States
SOURCE: Journal of Pediatrics, (1974) Vol. 84, No. 5, pp. 701-709.
CODEN: JOPDAB
DOCUMENT TYPE: Journal
FILE SEGMENT: 007 Pediatrics and Pediatric Surgery
005 General Pathology and Pathological Anatomy
022 Human Genetics
031 Arthritis and Rheumatism
LANGUAGE: English
AB The Winchester syndrome, a recently recognized inherited disorder of connective tissue, consists of dwarfism contractures, skin lesions, corneal opacities, osteoporosis, carpal tarsal osteolysis, and rheumatoid like small joint destruction. The third, fourth, and fifth recognized cases of this disorder are described. The features were progressive lysis of carpal and tarsal bones, replacement of bone and cartilage by dense fibrous tissue containing abnormal blood vessels, scanty trabecular and cortical bone, but normal resting cartilage and growth plates, hypervascularity apparently associated with osteolysis at large joints and widespread proliferation of ultrastructurally abnormal fibroblasts. Although this disorder was originally postulated to be a new mucopolysaccharidosis, no evidence was found for a lysosomal storage disease, and a reclassification of this disorder as a nonlysosomal connective tissue disease is proposed.

=> d his

(FILE 'HOME' ENTERED AT 18:12:21 ON 22 DEC 2006)

FILE 'CAPLUS, BIOSIS, MEDLINE, EMBASE' ENTERED AT 18:12:40 ON 22 DEC 2006
L1 1 S MEIKLE W?/IN
L2 73 MEIKLE
L3 0 L2 AND (SAPOSIN OR PROSAPOSIN)

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L4            94 HOPWOOD
L5             0 L3 AND (SAPOSIN OR PROSAPOSIN)
L6           453 WINCHESTER
L7             0 L6 AND (SAPOSIN OR PROSAPOSIN)
L8            73 L2
L9             0 L2 AND (LYSOSOME OR LYSOSOMAL)
L10            8 L4 AND (LSYOSOME OR LYSOSOMAL)
L11            8 L6 AND (LYSOSOME OR LYSOSOMAL)

=> gaucher or gaucher's
MISMATCHED QUOTE 'GAUCHER'S'
Quotation marks (or apostrophes) must be used in pairs,
one before and one after the expression you are setting
off or masking.

=> s (gaucher or gaucher's)
MISMATCHED QUOTE 'GAUCHER'S'
Quotation marks (or apostrophes) must be used in pairs,
one before and one after the expression you are setting
off or masking.

=> s (gaucher or "gaucher's" or gauchers)
L12       10751 (GAUCHER OR "GAUCHER'S" OR GAUCHERS)

=> L12 and (saposin or prosaposin or saposins or prosaposins)
L13         192 L12 AND (SAPOSIN OR PROSAPOSIN OR SAPOSINS OR PROSAPOSINS)
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EAST Search History

Ref #	Hits	Search Query	DBs	Default Operator	Plurals	Time Stamp
S1	71	saposin and (gaucher or gaucher's)	US-PGPUB; USPAT; DERWENT	OR	OFF	2006/12/22 16:51
S2	69	saposin and (gaucher or gaucher's) and (blood or serum or plasma)	US-PGPUB; USPAT; DERWENT	OR	OFF	2006/12/22 17:22
S3	76	saposin and (gaucher or gaucher's) and (blood or serum or plasma)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 17:23
S4	7	S3 not S2	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 17:22
S5	19	saposin and (gaucher or gaucher's) and (amniotic or urine)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 17:28
S6	124	saposin and (gaucher or gaucher's or lysosomal)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 17:28
S7	190	(saposin or prosaposin) and (gaucher or gaucher's or lysosomal)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:43
S8	104	(saposin or prosaposin) and (fabry's or fabry or fabrys or niemann-pick or pompe or pompes or pompe's or wolman)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:47
S9	97	S8 and (blood or serum or plasma or urine or amniotic)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:46
S10	190	(saposin or prosaposin) and (gaucher or gaucher's or lysosomal)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:46
S11	124	saposin and (gaucher or gaucher's or lysosomal)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:46
S12	76	saposin and (gaucher or gaucher's) and (blood or serum or plasma)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:46
S13	19	saposin and (gaucher or gaucher's) and (amniotic or urine)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:46
S14	69	saposin and (gaucher or gaucher's) and (blood or serum or plasma)	US-PGPUB; USPAT; DERWENT	OR	OFF	2006/12/22 18:46

EAST Search History

S15	7	S12 not S14	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:46
S16	71	saposin and (gaucher or gaucher's)	US-PGPUB; USPAT; DERWENT	OR	OFF	2006/12/22 18:46
S17	192	(saposin or prosaposin) and (fabry's or fabry or fabrys or niemann-pick or pompe or pompes or pompe's or wolman or lysosome or lysosomal)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:47
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S20	124	saposin and (gaucher or gaucher's or lysosomal)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:48
S21	76	saposin and (gaucher or gaucher's) and (blood or serum or plasma)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:48
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S23	69	saposin and (gaucher or gaucher's) and (blood or serum or plasma)	US-PGPUB; USPAT; DERWENT	OR	OFF	2006/12/22 18:48
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EAST Search History

S30	19	saposin and (gaucher or gaucher's) and (amniotic or urine)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:48
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S33	192	(saposin or prosaposin) and (fabry's or fabry or fabrys or niemann-pick or pompe or pompes or pompe's or wolman or lysosome or lysosomal)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:48
S34	124	S33 not S27	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:48
S35	104	(saposin or prosaposin) and (fabry's or fabry or fabrys or niemann-pick or pompe or pompes or pompe's or wolman)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:48
S36	97	S35 and (blood or serum or plasma or urine or amniotic)	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:48
S37	0	S34 not >"1999"	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:49
S38	0	S34 not ay>"1999"	US-PGPUB; USPAT; DERWENT	OR	ON	2006/12/22 18:49